



EIF2B5 gene

eukaryotic translation initiation factor 2B subunit epsilon

Normal Function

The *EIF2B5* gene provides instructions for making one of five parts of a protein called eIF2B, specifically the epsilon subunit of this protein. The eIF2B protein helps regulate overall protein production (synthesis) in the cell by interacting with another protein, eIF2. The eIF2 protein is called an initiation factor because it is involved in starting (initiating) protein synthesis.

Under some conditions, eIF2B increases protein synthesis by helping to recycle molecules called GTP, which carry energy to the initiation factor. Under other conditions, it slows protein synthesis by binding tightly to the initiation factor, which converts the eIF2B protein into an inactive form and prevents recycling of GTP.

Proper regulation of protein synthesis is vital for ensuring that the correct levels of protein are available for the cell to cope with changing conditions. For example, cells must synthesize protein much faster if they are multiplying than if they are in a resting state.

Health Conditions Related to Genetic Changes

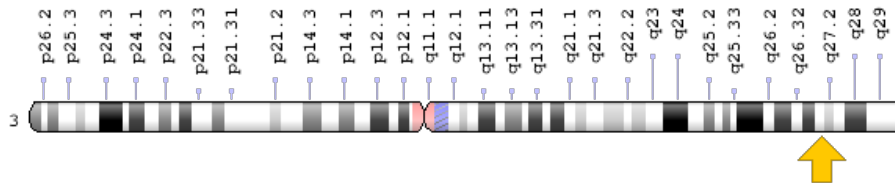
leukoencephalopathy with vanishing white matter

Mutations in the *EIF2B5* gene have been identified in about 65 percent of people with leukoencephalopathy with vanishing white matter, including those with a severe, early-onset form that is seen among the Cree and Chippewyan populations of Quebec and Manitoba (Cree leukoencephalopathy) and some affected females with a variant of the disorder in which the neurological features are accompanied by ovarian failure (ovarioleukodystrophy). These mutations cause partial loss of eIF2B function. Impairment of eIF2B function makes it more difficult for the body's cells to regulate protein synthesis and deal with changing conditions and stress. Researchers believe that cells in the white matter (nerve fibers covered by a fatty substance called myelin that insulates and protects nerves) may be particularly affected by an abnormal response to stress, resulting in the signs and symptoms of leukoencephalopathy with vanishing white matter.

Chromosomal Location

Cytogenetic Location: 3q27.1, which is the long (q) arm of chromosome 3 at position 27.1

Molecular Location: base pairs 184,134,435 to 184,145,311 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CACH
- CLE
- EI2BE_HUMAN
- EIF-2B
- eIF-2B GDP-GTP exchange factor
- EIF2Bepsilon
- eukaryotic translation initiation factor 2B, subunit 5 (epsilon, 82kD)
- eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa
- LVWM

Additional Information & Resources

Educational Resources

- Eureka Bioscience: Mechanism of Translation Initiation in Eukaryotes
<https://www.ncbi.nlm.nih.gov/books/NBK6597/>
- Molecular Biology of the Cell (fourth edition, 2002): The Phosphorylation of an Initiation Factor Globally Regulates Protein Synthesis
<https://www.ncbi.nlm.nih.gov/books/NBK26890/#A1387>

GeneReviews

- Childhood Ataxia with Central Nervous System Hypomelination/Vanishing White Matter
<https://www.ncbi.nlm.nih.gov/books/NBK1258>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28EIF2B5%5BTIAB%5D%29+OR+%28%28CACH%5BTIAB%5D%29+OR+%28CLE%5BTIAB%5D%29+OR+%28EIF-2B%5BTIAB%5D%29+OR+%28EIF2Bepsilon%5BTIAB%5D%29+OR+%28LVWM%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 5
<http://omim.org/entry/603945>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=EIF2B5%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3261
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8893>
- UniProt
<http://www.uniprot.org/uniprot/Q13144>

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Reviewed: October 2007
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
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